UVSSA gene

UV stimulated scaffold protein A

Normal Function

The *UVSSA* gene provides instructions for making a protein that is involved in repairing DNA damaged by ultraviolet (UV) rays from the sun. The damage can block vital cell activities such as gene transcription, which is the first step in protein production. If left uncorrected, DNA damage accumulates, which causes cells to malfunction and can lead to cell death.

Cells have several mechanisms to correct DNA damage. The UVSSA protein is involved in one mechanism that repairs damaged DNA within active genes (those genes undergoing gene transcription). When DNA in active genes is damaged, the enzyme that carries out gene transcription (RNA polymerase) gets stuck, and the process stalls. Researchers think that the UVSSA protein helps remove RNA polymerase from the damaged site, so the DNA can be repaired. Part of the UVSSA protein's role in this process is to ensure that another important protein called CSB is not broken down by exposure to UV rays.

Health Conditions Related to Genetic Changes

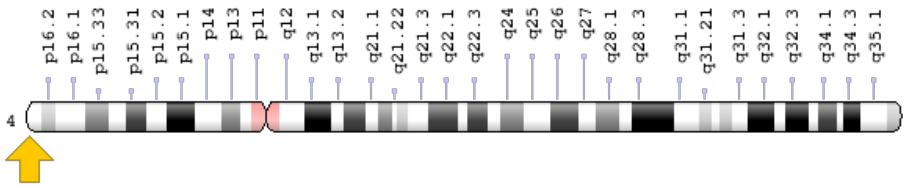
UV-sensitive syndrome

Mutations in the *UVSSA* gene cause UV-sensitive syndrome, which is a disorder characterized by sun sensitivity. People with this condition sunburn easily and have freckled skin or other changes in skin coloring (pigmentation). At least three *UVSSA* gene mutations have been identified, and these mutations eliminate the production of the UVSSA protein. Without this protein, skin cells cannot repair DNA damage caused by UV rays, and transcription of damaged genes is blocked. However, it is unclear exactly how a loss of the UVSSA protein causes the signs and symptoms of UV-sensitive syndrome.

Chromosomal Location

Cytogenetic Location: 4p16.3, which is the short (p) arm of chromosome 4 at position 16.3

Molecular Location: base pairs 1,345,662 to 1,388,049 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA1530
- UV-stimulated scaffold protein A
- UVSS3
- UVSSA_HUMAN

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): DNA Repair https://www.ncbi.nlm.nih.gov/books/NBK26879/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28UVSSA%5BTIAB%5D%29+OR+%28KIAA1530%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- UV-STIMULATED SCAFFOLD PROTEIN A http://omim.org/entry/614632

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_UVSSA.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=UVSSA%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29304
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/57654
- UniProt
  http://www.uniprot.org/uniprot/Q2YD98

Sources for This Summary

- Nakazawa Y, Sasaki K, Mitsutake N, Matsuse M, Shimada M, Nardo T, Takahashi Y, Ohyama K, Ito K, Mishima H, Nomura M, Kinoshita A, Ono S, Takenaka K, Masuyama R, Kudo T, Slor H, Utani A, Tateishi S, Yamashita S, Stefanini M, Lehmann AR, Yoshiura K, Ogi T. Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair. Nat Genet. 2012 May;44(5):586-92. doi: 10.1038/ng.2229.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22466610
- Schwertman P, Lagarou A, Dekkers DH, Raams A, van der Hoek AC, Laffeber C, Hoeijmakers JH, Demmers JA, Fousteri M, Vermeulen W, Marteijn JA. UV-sensitive syndrome protein UVSSA recruits USP7 to regulate transcription-coupled repair. Nat Genet. 2012 May;44(5):598-602. doi: 10.1038/ng.2230.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22466611
- OMIM: UV-STIMULATED SCAFFOLD PROTEIN A
  http://omim.org/entry/614632
- Zhang X, Horibata K, Saijo M, Ishigami C, Ukai A, Kanno S, Tahara H, Neilan EG, Honma M, Nohmi T, Yasui A, Tanaka K. Mutations in UVSSA cause UV-sensitive syndrome and destabilize ERCC6 in transcription-coupled DNA repair. Nat Genet. 2012 May;44(5):593-7. doi: 10.1038/ng.2228.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22466612

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/UVSSA

Reviewed: July 2012
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services